Medical Technologies Journal, Volume: 3, Issue: 4, October-December 2019, Pages: 485-486. Doi : https://doi.org/10.26415/2572-004X-vol3iss4p485-486

# Genotyping of *MEFV* and *SAA1* Genes and Their Correlation to the AA-Amyloidosis Development

**Type of article: conference abstract**

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**Abstract:**

**Background:** Familial Mediterranean fever (FMF) is the most common autoinflammatory disease caused by recessive mutations in the *MEFV* gene. If not treated, FMF patients may develop renal AA-amyloidosis that leads to renal failure and death. Both mutations and polymorphisms in *MEFV* and *SAA1* genes, respectively, have been associated with AA-amyloidosis in several populations. In Algeria, as FMF is still under-estimate and misdiagnosed, genetic data on renal complication are largely lacking. We thus explored the contribution of *MEFV* and *SAA1* loci in the development of amyloidosis in Algerian patients with FMF.

**Methods:** This study included 64 unrelated FMF patients (21 without and 43 with renal amyloidosis) and 13 healthy controls. The entire exon 10 was sequenced after PCR amplification to detect *MEFV* mutations. Genotypes of *SAA1* locus (SAA1.1, SAA1.5, and SAA1.3) were determined by PCR-RFLP (restriction fragment length polymorphism).

**Results:** Analyze of *MEFV* gene showed that the percentage of homozygous for p.M694I mutation was significantly higher in patients with amyloidosis compared to patients without amyloidosis (p=0.032). The SAA1.1/1.1 genotype was significantly predominant in patients with amyloidosis compared to those without AA-amyloidosis (p=0.001) and controls (0.001). The SAA1.5/1.5 genotype was identified only in patients without amyloidosis and controls. The most patients with renal complications were homozygous for p.M694I and SAA1.1 alleles.

**Conclusion:** Our data suggest a positive correlation between the p.M694I/M694I and SAA1.1/1.1 genotypes and the development of AA-amyloidosis secondary to FMF in Algerian patients.

**Keywords:** AA-Amyloidosis, Familial Mediterranean Fever, *MEFV* Gene, *SAA1* Polymorphisms.

## 1. Declaration of conflicts

This article is a conference abstract selected from the abstract book of the International Congress on Health Sciences and Medical Technologies ICHSMT'19, Tlemcen, Algeria, December 05-07, 2019.

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## 2. Authors' Biography

No biography

## 3. References

No references